Genetic testing currently validated at ICDL as of January 2016

Metaphase cytogenetics based testing:

Amniotic Fluid
Chorionic Villus samples
Bone marrows
Leukemic Bloods
Constitutional bloods
Products of conception
Solid tumors (all types)
Skin biopsies for mosaicism

Chromosomal SNP Microarray

Constitutional testing
Neoplastic testing for patients with MDS, B- and T-cell ALL and select solid tumor types.

FISH probes validated for metaphase or interphase analysis:

Constructs: BAP=breakapart, DCDF= dual color dual fusion, All other probes are for enumeration

Constitutional FISH (enumeration):

- ELN/D7Z1; Williams Syndrome
- SNRPN1; Prader-Willi syndrome
- TBX1/N85A3; DiGeorge/Velocardiofacial Syndrome and 22q13 microdeletion syndrome
- XIST; X linked inactivation center
- CEPX/CEPY; gender determination and post bone marrow transplant engraftment testing

Any of the following probes can be run as part of a panel or as a single test, depending on the clinical situation of the patient and whether the analysis is at diagnosis or being used for monitoring purposes

BAP = Breakapart
DCDF = dual color, dual fusion
All other probes for enumeration

- CEPX/CEPY post gender-mis-match bone marrow transplant engraftment testing

Chronic Lymphocytic Leukemia

- TP53/ATM (deletion)
- D13S319/CEP12 (deletion)/(Trisomy)
- CDKN2A/B (associated with Richter's Transformation)
- CCND1/IGH (DCDF) to r/o MCL

Myelodysplastic Syndrome and/or Acute Myelogenous Leukemia

- EGR1/D5S721 (5q deletion)
- D7S486/CEP7 (7q deletion/monosomy 7)
- CEP8/D20S108 (Trisomy 8/20q deletion)
- ETV6/RUNX1 (12p and 21 del or rea)
- CBFB (BAP) (inv(16))
- PML/RARA (DCDF) (t(15;17))
- KMT2A (BAP) (translocations involving 11q23, amplification, deletion)
- EVI1 (BAP) (inv(3)(q) or t(3q))
- RUNX1/RUNX1T1 (DCDF) (t(8;21))
- RARA (BAP) (assess for alternate RARA gene rearrangements)

Myeloproliferative Neoplasms

- BCR/ABL1 (DCDF) t(9;22)
- PDGFRa (BAP) 4q21
- PDGFrb (BAP) 5q33
- FGFR1 (BAP) 8p11

Plasma Cell Neoplasms/Amyloidosis (CD138+ enriched)

Standard Panel:

- 1p36/1q25 (deletion of 1p or gain of 1q)
- KMT2A (gain of chromosome 11/11q)
- TP53/D17Z1 (17p deletion)
- IGH (BAP) (to distinguish between Trisomy 14 and IGH gene rearrangement)
- MYC (BAP)

With further reflex to:

- CEP 7,9,15 to assess for hyperdiploidy
- CCND1/IGH (DCDF) t(11;14)
- FGFR3/IGH (DCDF) t(4;14)
- MYEOV/IGH (DCDF) t(11;14)
- MAF/IGH (DCDF) t(14;16)
- MAFB/IGH (DCDF) t:14;20)
- MYC/IGH (DCDF) t(8;14)

Lymphoma:

- BCL6 (BAP) (translocations involving 3q26, DLBCL)
- MYC (BAP) (translocations or gene amplification)
- MYC/IGH/CEP8 (DCDF) (translocations or gene amplification)
- BCL2/IGH (DCDF) (t(14;18) or variants, including BCL2 gene amplification)
- CCND1/IGH (DCDF) (t(11;14) or variants)
- CDKN2A (9p deletion)
- IGK
- IGL
- ALK

Acute Lymphocytic leukemia (Pediatric)

- CEP4/CEP10/CEP17 assessment for hyperdiploidy (Trisomy assessment)
- BCR/ABL1 (DCDF) (t(9;22)
- CDKN2A/D9Z3 (deletion of 9p)
- ETV6/RUNX1 (DCDF) t(12;21)
- KMT2A(BAP) (translocations involving 11q23)
- ETV6 (BAP) (alternate ETV6 translocations versus Trisomy 12)
- PDGFRb
- PAX5
- IGH (BAP)
- IKZF1
- TCF3 (BAP)

Acute Lymphocytic Leukemia (Adult)

- BCR/ABL1
- MYC/IGH
- PDGFRb
- CDKN2A
- ETV6/RUNX1
- IGH (BAP)

Breast Carcinoma:

- ERBB2(HER2) (gene amplification, monosomy, polysomy, genetic heterogeneity in BrCa)

Solid tumors (touch prep or from fixed cell pellet):

- PAX3, PAX7; Rhabdomyosarcoma
- MYCN, 1p, 11q; Neuroblastoma
- MYC and MYCN; Medulloblastoma, Angiosarcoma